Table 1. Common Congenital Muscular Dystrophies

Disease Entity	Protein Product (*Gene Symbol*)	Clinical Features
Congenital muscular dystrophy with primary laminin 2 (merosin) deficiency (MDC1A)	Laminin α2 (*LAMA2*)	Sitting and standing with support as maximal motor ability; neuropathy; epilepsy in approximately 30%; possible subclinical cardiomyopathy; generally normal mental development
Congenital muscular dystrophy with partial merosin deficiency (MDC1B)	Not known	Rare; variety of severity; delayed onset possible; limb girdle weakness; generalized muscle hypertrophy; early respiratory failure possible
Fukutin-related proteinopathy (MDC1C)	Fukutin-related protein (*FKRP*)	Often reminiscent of MDC1A but severity more variable; generally normal mental development; structural brain involvement and mental retardation possible
LARGE-related congenital muscular dystrophy (MDC1D)	Acetylglucosaminyltransferase-like protein (*LARGE*)	Congenital muscular dystrophy with profound mental retardation can eventually blend with the muscle–eye–brain disease/Walker-Warburg syndrome spectrum
Fukuyama congenital muscular dystrophy	Fukutin (*FCMD*)	Frequent in Japanese population; never walk; mental retardation; epilepsy common—clinical overlap with muscle–eye–brain disease
Muscle–eye–brain disease	Protein-O-linked mannose β1, 2-N-acetylglucosaminyl-tranferase 1 (*POMGnT1*), also caused by FKRP, FCMD	Severe weakness and mental retardation; large head; prominent forehead; flat midface; walking rarely achieved; ocular involvement (eg, severe myopia, retinal hypoplasia); motor deterioration because of spasticity
Walker-Warburg syndrome	O-mannosyltranferase 1 (*POMT1*), also *POMT2*, *FKRP*, *FCMD*	Severe; lethal within first years of life because of severe central nervous system involvement
Ullrich congenital muscular dystrophy and Bethlem myopathy	$\alpha_{1/2}$ and α_3 collagen VI (*COL6A1*, *COL6A2*, *COL6A3*)	Distal joint hyperextensibility; proximal contractures; motor abilities variable; precludes independent ambulation in severe Ulrich cases; soft palmar skin
Integrin α7	Integrin α7 (*ITGA7*)	Very rare; delayed motor milestones; walking within 2 to 3 years of life
Rigid spine muscular dystrophy	Selenoprotein N (*SEPN1*)	Delayed walking; predominantly axial weakness with early development of spine rigidity; restrictive respiratory syndrome
Lamin A/C-related congenital muscular dystrophy	Lamin A/C (*LMNA*)	Early motor deterioration; prominent axial weakness with dropped head syndrome; early development of spinal rigidity